Chanarin-Dorfman syndrome

Chanarin-Dorfman syndrome is a condition in which fats (lipids) are stored abnormally in the body. Affected individuals cannot break down certain fats called triglycerides, and these fats accumulate in organs and tissues, including skin, liver, muscles, intestine, eyes, and ears. People with this condition also have dry, scaly skin (ichthyosis), which is usually present at birth. Additional features of this condition include an enlarged liver (hepatomegaly), clouding of the lens of the eyes (cataracts), difficulty with coordinating movements (ataxia), hearing loss, short stature, muscle weakness (myopathy), involuntary movement of the eyes (nystagmus), and mild intellectual disability.

The signs and symptoms vary greatly among individuals with Chanarin-Dorfman syndrome. Some people may have ichthyosis only, while others may have problems affecting many areas of the body.

Frequency

Chanarin-Dorfman syndrome is a rare condition; its incidence is unknown.

Genetic Changes

Mutations in the *ABHD5* gene cause Chanarin-Dorfman syndrome. The *ABHD5* gene provides instructions for making a protein that turns on (activates) the ATGL enzyme, which breaks down triglycerides. Triglycerides are the main source of stored energy in cells. These fats must be broken down into simpler molecules called fatty acids before they can be used for energy.

ABHD5 gene mutations impair the protein's ability to activate the ATGL enzyme. An inactive enzyme makes the breakdown of triglycerides impossible, causing them to accumulate in tissues throughout the body. The buildup of triglycerides results in the signs and symptoms of Chanarin-Dorfman syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- CDS
- Chanarin-Dorfman disease

- Ichthyotic neutral lipid storage disease
- neutral lipid storage disease with ichthyosis
- Triglyceride storage disease with ichthyosis
- triglyceride storage disease with impaired long-chain fatty acid oxidation

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Triglyceride storage disease with ichthyosis
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268238/

Other Diagnosis and Management Resources

- MedlinePlus Encyclopedia: Ichthyosis vulgaris
  https://medlineplus.gov/ency/article/001451.htm

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Ichthyosis vulgaris
  https://medlineplus.gov/ency/article/001451.htm
- Health Topic: Cataract
  https://medlineplus.gov/cataract.html
- Health Topic: Lipid Metabolism Disorders
  https://medlineplus.gov/lipidmetabolismdisorders.html

Genetic and Rare Diseases Information Center

- Chanarin-Dorfman syndrome
  https://rarediseases.info.nih.gov/diseases/3979/chanarin-dorfman-syndrome

Additional NIH Resources

- National Eye Institute: Facts About Cataract
  https://nei.nih.gov/health/cataract/cataract_facts
- National Institute of Neurological Disorders and Stroke: Lipid Storage Diseases Information Page
  https://www.ninds.nih.gov/Disorders/All-Disorders/Lipid-storage-diseases-Information-Page

Educational Resources

- Disease InfoSearch: Chanarin-Dorfman Syndrome
  http://www.diseaseinfosearch.org/Chanarin-Dorfman+Syndrome/1269
- MalaCards: chanarin-dorfman syndrome
  http://www.malacards.org/card/chanarin_dorfman_syndrome
- Orphanet: Neutral lipid storage disease
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=165

Patient Support and Advocacy Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST)
  http://www.firstskinfoundation.org/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22chanarin-dorfman+syndrome%22+OR+%22lipid+metabolism%2C+inborn+errors%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28chanarin-dorfman+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CHANARIN-DORFMAN SYNDROME
  http://omim.org/entry/275630

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